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Mainstreaming genomics in the National Health Service in England: a survey to understand preparedness and confidence among paediatricians

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ABSTRACT

Background The National Health Service in the UK is the first national healthcare system to offer genomic sequencing for rare disease diagnosis as routine care. Non-genetic medical specialists, including paediatricians, can now request genomic testing for certain clinical indications. The primary purpose of this study was to evaluate the preparedness and confidence of paediatricians providing genomic sequencing in England. In addition, we assessed current practice, perceived utility of testing, barriers and enablers, prior genomics education and training preferences.

Methods A 26-item electronic survey for completion by paediatric specialists. Participants were recruited through national associations and a conference. Quantitative items were analysed using descriptive and inferential statistics. Open-ended question responses were analysed by qualitative content analysis.

Results 157 responses were included in the analysis. Only 49.0% reported feeling prepared for mainstreaming despite 75.0% reporting they had requested testing in the past 12 months, 47.7% indicating they had returned genomic sequencing results and 67.1% feeling genomic testing was useful. Mean confidence scores were lowest for tasks including using human phenotype ontology terminology on test request forms (3.9/10), interpreting genomic test results (4.8/10), discussing complex genomic results with patients and families (4.3/10) and integrating test results into patient care (4.7/10). Significantly higher average ranked genomic confidence was identified among those who had requested testing in the last 12 months compared with those who had not ($Z=5.063$, $p<0.001$, $r=0.412$). The most frequent barriers to mainstreaming were lack of training and knowledge (43.3%), determining patient eligibility (28.0%), lack of time (27.4%) and confidence (25.5%). Webinars (48.4%), followed by continued professional development meetings and/or conferences (38.9%), were the preferred mode of training.

Conclusions Our data suggest that preparedness and confidence among paediatricians in genomics is currently lacking. Support from clinical genetics services, simplified referral forms and webinar training sessions could improve current practice.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Many non-genetic clinicians do not feel adequately prepared to provide patients with genomic testing; however, to date, no studies have explored preparedness and confidence among paediatricians in England who are likely to see patients who may benefit from genomic testing.

WHAT THIS STUDY ADDS

⇒ Our findings suggest that paediatricians are providing genomic services despite not yet feeling sufficiently prepared and confident.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ Our findings highlight that education and support from clinical genetics colleagues would improve paediatricians' confidence in completing genomic tasks. Simplified referral paperwork and clearer pathways for onward management would also potentially remove some of the current barriers.

INTRODUCTION

Genomic sequencing tests include gene panels in which a phenotypically targeted selection of genes is sequenced and analysed, whole genome sequencing (WGS) in which the entire genome is sequenced, and whole exome sequencing where the coding regions of the genome are fully sequenced. These have been demonstrated to provide a higher diagnostic yield in shorter time frames among paediatric patients than targeted single gene and microarray tests.[1–3] Achieving a genetic diagnosis may target care more effectively, avoid unhelpful interventions, provide prognostic information, support reproductive planning and enable increased peer and social support for families affected by rare diseases.[4 5]

The UK National Health Service (NHS) is the first national healthcare system in the world to offer WGS for rare disease diagnosis (as well as cancer) as part of routine care.[6 7] Within England, genomics services are organised into seven genomic laboratory hubs, commissioned to deliver the Genomic

Medicine Service (GMS) since 2018.[7] The National Genomic Test Directory documents the genomic tests available, detailing the appropriate test that should be delivered for each clinical indication, eligibility criteria and appropriate referral specialisms. It includes the provision of WGS for many indications and in most cases, genomic testing can be requested by non-genetic medical specialists (known as 'mainstreaming'). Mainstreaming has been necessitated by the expansion of available genomic tests, increased clinical utility, the incorporation of genomic tests into standard clinical care pathways and the limited resources of clinical genetics services. Since July 2021, paediatric specialties have been able to order WGS for indications such as developmental delay and intellectual disability.[8]

Despite the benefits of mainstreaming, research has shown that many non-genetic clinicians do not feel adequately prepared and confident to provide patients with genomic tests.[9–12] Few studies in the UK have explored the preparedness and confidence of non-genetic clinicians for genomic medicine. A workforce-wide needs analysis of 2814 individuals representing 10 workforce groups found that while 31.3% of responders indicated they were currently involved in the delivery of genetic and/or genomic services, of those, 19.8% had no previous genomics education and training.[13] Overall, between 75.9% and 85.7% indicated that they felt they needed further training in genomics. A nationwide survey of the genomics training needs of UK oncologists, completed by 150 oncologists (representing 10% of UK oncologists) found that formal training in genomics had not been received by 38.7% of oncologists and 92.7% identified a need for additional genomics training.[14] A survey of 100 gastroenterology trainees reported that only 9% felt their training had prepared them sufficiently to incorporate genomic medicine in their clinical practice.[11] An evaluation of a Master's programme in Genomic Medicine found that learners were a diverse cohort of healthcare professionals (HCPs) including doctors, healthcare scientists, nurses and midwives.[15] Both learners and their managers reported enhanced genomic practice after completing their studies. An interview study, primarily with community paediatricians working in England, revealed a lack of familiarity with the new processes for requesting genomic tests, impacting confidence.[16] Most of those interviewed had attended educational sessions but had found making time for training challenging. Participants suggested that the test request process required streamlining, and improvements were needed in information sharing and access to test records. To our knowledge, that is the only study exploring the experience of paediatricians using the GMS which has been conducted in the UK.

Examining the views of paediatricians is important because many genomic conditions commonly present in childhood, including congenital structural malformations and neurodevelopmental delay. Paediatricians will, therefore, have a key role in facilitating genomic tests for their patients including seeking informed consent, understanding and explaining genomic test reports, managing a genomic diagnosis as well as knowing when it is appropriate to refer patients on to clinical genetics. Given the paucity of research exploring the experience of paediatricians, and the importance of mainstreaming in NHS England's agenda,[7] we conducted a survey with the primary purpose of understanding and quantifying how prepared and confident paediatricians feel for integrating genomic testing into their clinical practice. In addition, we assessed current practice and familiarity with tasks involved in genomic testing, the perceived utility of testing, barriers and enablers to providing genomic testing, prior genomics education and training preferences.

METHODS

Study design

This study forms part of a wider mixed-methods evaluation of the NHS GMS for paediatric rare diseases.[17] An anonymous online quantitative cross-sectional survey targeted at paediatricians working in the NHS in England was designed and promoted through the social media channels and E-newsletters of the British Association of Child Health, the British Association of Childhood Disability and through the Royal College of Paediatrics and Child Health annual conference as well as a regional Genomics Teaching Day for mainstream clinicians, 'The Great Genome Event'. The promotional information included an electronic link to the survey. Our study employed a convenience sampling approach and was exploratory in nature. As such, a formal sample size calculation was not undertaken.

Patient and public involvement

This mixed-methods evaluation has been designed with support from an advisory group including patient advocates from two genetics patient organisations. Specifically for this study, prior to designing the survey, RG discussed the study at an advisory meeting where it was agreed we would focus on the constructs 'preparedness' and 'confidence' because of the recent implementation of the mainstreaming agenda. Following data collection and analysis, RG presented the key findings from the survey, and members commented on implications for policy and practice.

Survey development

Questions around preparedness, confidence as well as education and training preferences (so that recommendations could be made) were identified and adapted for use from previous surveys in Australia[9 18 19] and the UK.[20] Questions were also sourced from a UK survey by Hill *et al* (manuscript in progress), which was part of a mixed-methods exploration of clinicians' opinions of the NHS rapid sequencing service for critically ill children. Questions were adapted to fit the context of this survey while

preserving the prior established meaning and readability.[21] Some additional survey questions were purposely designed by the authors. These included questions to explore: current practice, to understand how many responders were currently offering genomic testing; and questions around clinical utility to add context around paediatricians' preparedness and confidence. To complement constructs explored with other specialties in the wider mixed-methods evaluation of the GMS[17], questions also examined paediatricians' perceived barriers and enablers.

An extensive list of survey questions was assessed for usefulness, readability and answerability through consultation between RG, CL and two social science researchers working in genomics. Following this phase, some questions were revised and others excluded. A subsequent draft of the survey was then piloted in hour-long 'think-aloud' interviews with two paediatricians and a genetic counsellor. The survey was modified based on their feedback.

The final survey comprised 26 questions (online supplemental information I) including multiple-choice questions, Likert scales as well as space for free-text comments. We also collected demographic data including participant age range, gender, clinical specialty, clinical role, geographical location and years of experience. The survey took approximately 10 minutes to complete.

Data collection

The survey was made accessible online through a web-based electronic data capture software platform, REDCap.[22] Inclusion criteria were paediatric specialists working in the NHS in England. The survey was available from 1 February 2024 and closed on 1 April 2024. Consent was considered implied if participants chose to complete and submit the survey; however, no questions were mandatory. The anonymous nature of the survey was highlighted in the participant information sheet whereby no contact details were collected, and the estimation of time taken to complete the survey was provided. Completion of the survey was entirely voluntary and no incentives were offered to participants.

Data analysis

Data analysis was completed using the IBM SPSS Statistical Package V.27 to calculate descriptive statistics; frequencies, proportions, means and SD, and inferential statistics to make between-group comparisons.[23] The total participant number was adjusted so that it reflected the total number of participants responding to each survey question. Not all participants answered all survey questions. Questions were not filtered using question logic. Therefore, in some cases, participants responded 'no' to a question but were able to answer a subsequent question as if they had responded 'yes'. We present the data as they were received.

Confidence ratings for individual genomic tasks were consolidated as a mean to give an overall genomic confidence score. The Mann-Whitney test for comparison of two variables and the Kruskal-Wallis test for three variables and more were used to identify variables associated with differences in overall genomic confidence. Genomic confidence was compared between individuals grouped by frequency of test ordering, familiarity with genomic testing and preparedness for genomic practice. A p value of 0.05 (two-sided) was considered significant. Significance values were adjusted by the Bonferroni correction for multiple tests. Effect size (r) was calculated where significant differences were identified by Dunn's post hoc pairwise comparison. 0.1 indicated a small effect size, 0.3 a medium effect size and 0.5 a large effect size.[24] Surveys were excluded if only demographic data were provided. The number of missing items was reported.

Inductive content analysis was employed to inductively identify the content categories present and their frequency in the free-text responses.[25]

RESULTS

Study sample and demographics

A total of 165 completed survey responses were recorded (none were excluded due to missing data). Of those, eight participants were excluded as they worked outside England (Wales n=6 and Northern Ireland n=2) leaving a total of 157 for analysis. Due to the recruitment methods used, it was not possible to know the number of paediatricians the survey invite reached, nor the number who started but did not submit the survey. Therefore, an accurate response rate could not be determined. The characteristics of the study sample are described in table 1. Participants represented all age groups, years of experience and GMS locations with the largest proportion of participants being female hospital-based consultants.

Current practice

Ordering genomic tests

Over half of participants (59.6%, n=93) said that they regularly offer genetic and/or genomic tests in their clinical practice (table 1). In total, 117 (75.0%, n=156) participants reported having ordered genomic sequencing tests in the previous 12 months. Most frequently, tests were requested once or twice a year (36.8%, n=43/117), followed by monthly (26.5%, n=31/117) and quarterly (23.1%, n=27/117). One participant reported requesting testing daily (0.9%), 6 weekly (5.1%) and 9 fortnightly (7.7%). Of those participants who indicated they had not ordered genomic testing for a patient in the last 12 months (32.5%, n=51/157), the most common reason selected was that they referred eligible patients to a specialist team (35.3%, n=18/51). Just over one-quarter (25.5%, n=13/51) cited reasons relating to perceived relevance to practice, that is, they were unsure of the relevance of testing to their practice (9.8%, n=5/51) or did not feel genomic testing was relevant to their practice (15.7%, n=8/51). The remaining reasons were of a practical nature; 15.7% (n=8/51) were not permitted

Table 1 Participant characteristics and genetic/genomic testing experience

Variable	Number of participants (n)	%
Gender (n=157)		
Female	122	77.7
Male	33	21.0
Prefer not to answer	2	1.3
Age range (n=157)		
24 or under	1	0.6
25–34	42	26.8
35–44	50	31.8
45–54	48	30.6
55–64	14	8.9
65 or over	2	1.3
Clinical role (n=153)		
Consultant	74	48.4
Specialty registrar	51	33.3
Specialty doctor	16	10.5
Associate specialist doctor	3	2.0
Foundation doctor	4	2.6
Other	5	3.3
Working location (n=156)		
Hospital	126	80.8
Community	30	19.2
Years of experience (n=157)		
<1 year	5	3.2
1–5 years	39	24.8
6–10 years	30	19.1
11–15 years	31	19.7
16–20 years	16	10.2
>20 years	36	22.9
GMS working location (n=157)		
Central and South	32	20.4
North East and Yorkshire	32	20.4
South East	24	15.3
East	22	14.0
North West	18	11.5
North Thames	13	8.3
South West	10	6.4
Unsure	6	3.8
Current practice—offering genetic and/or genomic testing (n=156)		
I regularly offer genetic and/or genomic tests in my clinical practice	93	59.6
I am familiar with genetic and/or genomic tests but I do not personally offer them	40	25.6
I have heard of genetic and/or genomic tests but I am not very familiar with them	23	14.7
I have never heard of genetic and/or genomic tests	0	0

GMS, Genomic Medicine Service.

to request testing, and 7.8% (n=4/51) reported being unsure how to request testing. Of the eight individuals who selected 'other', reasons given were also of a practical nature and included that the consultant requested the testing (n=4), they had not yet had the opportunity (n=3) and the referral for testing had usually already been made before the patient reached them (n=1).

Familiarity with tasks involved in genomic testing

Participants were given a list of tasks associated with providing genomic sequencing testing for patients and asked to select which tasks they had completed (table 2). Most frequently, participants had discussed genomic testing with patients and their families (70.1%, n=110), organised to obtain and send blood samples for testing (69.4%, n=109), detailed relevant clinical information on test order forms (66.2%, n=104) and referred patients and their families on to clinical genetic specialists (61.1%, n=96). Least frequently, participants had integrated genomic sequencing results into the care of patients and their families (20.4%, n=32), discussed more complex results with patients and their families (25.5%, n=40) and selected suitable human phenotype ontology (HPO) terminology on test order forms (25.5%, n=40).

Returning genomic test results

Participants were asked if they had given genomic sequencing results to patients and/or their families (table 3). In total, just under half (74/155, 47.7%) responded that they had, with the largest proportion indicating that they did so independently, that is, without advice from a multidisciplinary team (MDT) or assistance from clinical genetics (n=36/76, 47.4%). Just over half (n=81/155, 52.3%) had not returned genomic results. Of the 79 who responded as to why, the most frequently cited reason was because they had not yet received any genomic test results (n=28/79, 35.4%) however, eight of these participants had responded that they had not ordered genomic sequencing tests in the past 12 months. Therefore, 20 participants (25.3%) who had ordered tests in the past 12 months had not given any results as they had not yet received any. Participants most frequently reported giving results to patients every 3–4 months (table 3).

Table 2 Familiarity with tasks involved in delivering genomic sequencing tests

Genetic task performed	n (157)	%
Considering whether or not a patient is eligible for testing	89	56.7
Liaising with the testing laboratory about patient eligibility	61	38.9
Discussing the test with patients and their families	110	70.1
Consenting patients and their families for the test	97	61.8
Detailing relevant clinical information on test order forms	104	66.2
Selecting suitable human phenotype ontology terminology on test order forms	40	25.5
Organising to obtain and send blood samples for testing	109	69.4
Interpreting the results report	48	30.6
Accessing resources to help if you have questions or concerns about results	42	26.8
Discussing results at multidisciplinary team meetings	52	33.1
Discussing straightforward results with patients and their families	81	51.6
Discussing more complex results with patients and their families, such as variants of uncertain significance or unexpected (incidental) findings unrelated to the reason for requesting the test	40	25.5
Integrating genomic sequencing test results into the care of patients and their families	32	20.4
Referring patients and their families on to clinical genetic specialists	96	61.1
Signposting patients and their families to other organisations	60	38.2
None of these tasks are applicable to my role	8	5.1

Table 3 Experience returning genomic test results

Survey question and response options	n	%
Have you given genomic sequencing test results to patients and or their families? (n=155)		
No	81	52.3
Yes	74	47.7
If yes, how does this more frequently happen? (n=76)		
Independently	36	47.4
With advice/support from an MDT	22	28.9
With assistance from Clinical Genetics	16	21.1
Other*	2	2.6
If no, why haven't you? (n=79)		
I have asked another healthcare professional to deliver the results	30	37.9
I haven't yet received any genomic test results	28	35.4
I haven't requested any genomic tests	21	26.6
How frequently do you return genomic sequencing results? (n=80)		
Weekly	2	2.5
Fortnightly	2	2.5
Monthly	11	13.8
Every 3–4 months	23	28.8
6 monthly	16	20.0
Annually	9	11.3
Less than once per year	17	21.3

*No reasons were given.
MDT, multidisciplinary team.

Use of information resources

Participants were asked if they use information resources designed for patients when discussing genomic sequencing testing with them. In total, 133 out of 157 participants responded to this question. Of those, 48.1% (n=64/133) indicated that they do. 26 participants specified a resource and of those, 11 stated that they used resources provided by support groups/charities, 7 used leaflets and resources from their local genetic services and three used resources from Genomics England. More general resources like Google searches and websites were also specified, and one participant suggested that they would like more resources (see online supplemental information II).

Preparedness and confidence

Preparedness for genomic testing

Almost half of participants (49.0%, n=76/155) either agreed (34.8%, n=54/155) or strongly agreed (14.2%, n=22/155) that they feel prepared to use genomic testing in their practice. Just over a quarter (28.4%, n=44/155) disagreed (23.2%, n=36/155) or strongly disagreed (5.2%, n=8/155) and 35 neither agreed nor disagreed (22.6%).

Confidence with genomic testing

Participants were asked to rank their confidence on a scale from 1 to 10 (1=not at all confident; 10=very confident), in completing each of the tasks. The mean confidence for each task was determined (figure 1). Participants felt most confident with referring patients on to clinical genetics (mean=7.1), organising blood samples (mean=7.0) and discussing straightforward results with families (mean=6.5). They felt least confident using HPO terminology in test request forms (mean=3.9), discussing complex results with families (mean=4.3) and integrating results into patient care (mean=4.7).

For each participant, the mean was determined across all the genomic tasks to give an overall genomic confidence score. Significantly higher average rank genomic confidence was identified among those who had requested testing in the last 12 months compared with those who had not (Z=5.063, p<0.001, r=0.412), with

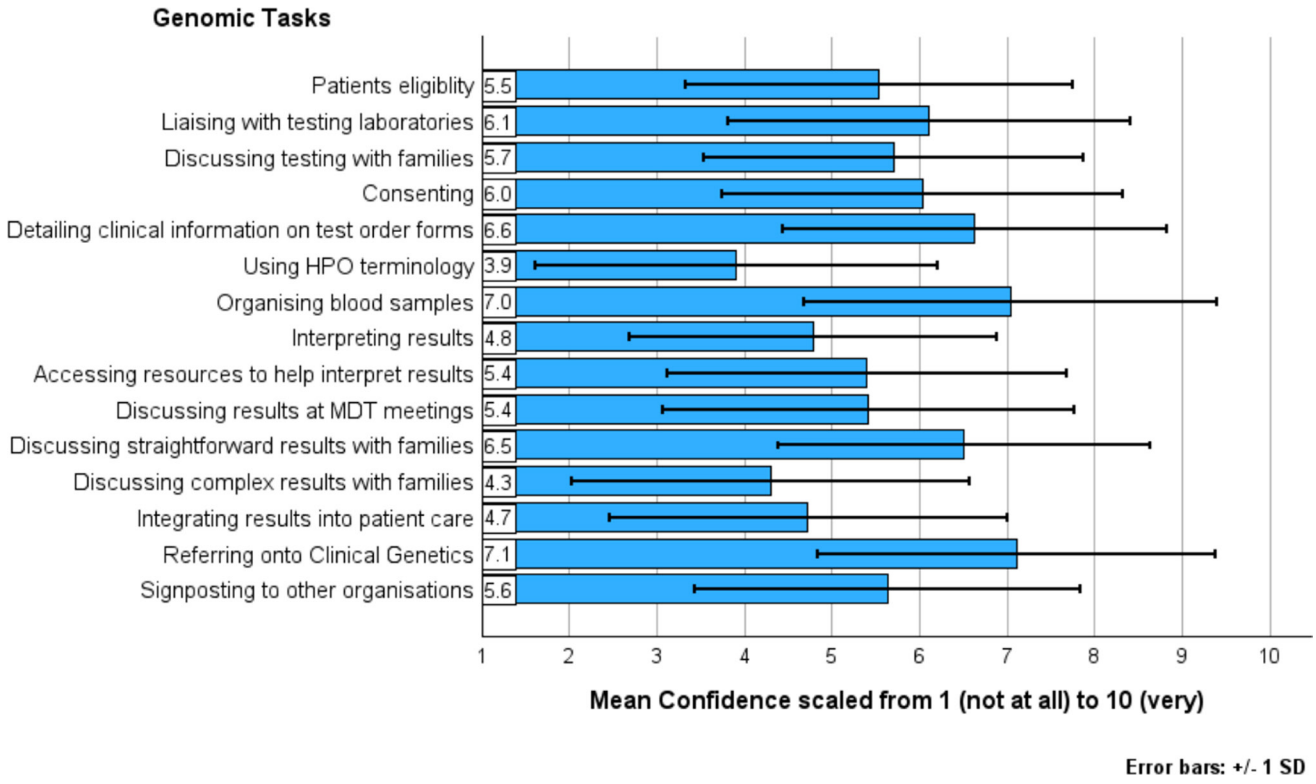

Figure 1 Confidence with mainstreaming genomic testing. HPO, human phenotype ontology; MDT, multidisciplinary team.

those referring fortnightly or monthly having significantly higher average ranked genomic confidence scores than those referring only once or twice per year (p=0.039, r=0.418 and p=0.028, r=0.364, respectively).

There were significant differences in genomic confidence scores among the participant groups with different reported familiarity with genomic testing (H (2)=47.640, p<0.001). Average ranked genomic confidence was significantly higher among those that regularly offer testing compared with those that have heard of genomic tests but are not very familiar (p<0.001, r=0.586) and those that are familiar with genomic testing but do not personally offer it (p<0.001, r=0.367). Additionally, participant preparedness was associated with significant differences in genomic confidence scores (H (2)=47.640, p<0.001) with ranked average confidence highest among those participants who agreed or strongly agreed that they feel prepared to use genomic testing.

Improving genomic confidence

When asked to select one option from a list that would improve their confidence most in completing genomic tasks, of 153 participants who responded to this question, 56 selected further genomic education (36.6%), 48 selected additional support from clinical genetics (31.4%), 33 selected experience (21.6%) and 13 selected support from an experienced colleague (8.5%). Three individuals selected 'other' (1.9%). Reasons specified included 'all of the above', 'help with administration' and 'video (for patients) to explain tests and request consent online'.

Views, experiences and preferences

Perceived utility of genomic testing

Regarding perceived utility of genomic testing in the past year in directing the management of patients with a suspected monogenic condition, the majority (67.1%, n=102/152) ranked genomic tests useful (42.8%, n=65/152) or very useful (24.3%, n=37/152). Only 4.0% (n=6/152) felt they were not (2.6%, n=4/152) or not at all useful (1.3%, n=2/152) and 28.9% (n=44/152) had found tests neither useful nor not useful.

Engagement with clinical genetics

Participants were asked if they had contacted clinical genetics in the past 12 months. Overall, 74.2% (n=115/155) responded that they had. Of those who had, the most commonly selected reason was for advice on the type of genomic test to order (70.4%, n=81/115), followed by for information about a suspected genetic condition (57.4%, n=66/115). 46 participants (40.0%) contacted clinical genetics for advice interpreting results, 41 (35.7%) for assistance with post-test genetic counselling, 27 (17.2%) for assistance with pretest counselling and for advice on how to refer a patient to clinical genetics, 21 (18.3%) for advice on completing test request forms, 14 (12.2%) for advice on taking informed consent and 13 (11.3%) on providing patient information such as leaflets. Two participants (1.7%) selected the option 'other', providing the reasons support with manpower for completing paperwork (n=1) and to check whether the laboratory had received a sample (n=1).

Table 4 Barriers and enablers to requesting genomic tests

	n (157)	%
Barriers		
Lack of training and knowledge	68	43.3
Insufficient time	46	29.3
Determining patient suitability/eligibility	44	28.0
Confidence	40	25.5
Selecting appropriate testing	37	23.6
Completing test request forms	30	19.1
Taking patient consent	21	13.4
Pretest counselling	19	12.1
Not part of my job role	18	11.5
Obtaining patient samples	16	10.2
Concern about delivering results to patients	12	7.6
Lack of patient benefits	6	3.8
Patient motivation	4	2.5
None of the above	8	5.1
Other*	3	1.9
Enablers		
Clinical genetics support	84	53.5
Simplified referral paperwork	78	49.7
Clearer pathways for onward management	70	44.6
Further genomics education	48	30.6
MDT meetings	41	26.1
Specialist roles for patient consent	33	21.0
Further on-the-job training	32	20.4
Patient information resources in different languages	30	19.1
Administrative support	26	16.6
Departmental genomic champions	25	15.9
Greater departmental support	15	9.6
None of the above	1	0.6
Other†	5	3.2

*Other barriers included: insufficient funding for teams (1/3), long test turn- around times (1/3) and being unable to complete this work without the support of genomic practitioners (1/3).
†No enablers were specified.
MDT, multidisciplinary team.

Barriers and enablers to providing genomic testing

Participants were asked to select their top three barriers to requesting genomic sequencing tests for patients from a list of options (table 4). The most frequently cited barriers were lack of training and knowledge (43.3%, n=68/157), determining patient suitability/eligibility (28.0%, n=44/157), insufficient time (27.4%, n=43/157) and confidence (25.5%, n=40/157).

Participants were asked to select three options from a list of possible enablers that would help support them in offering genomics testing to patients (table 4). The most frequently cited responses were clinical genetics support (53.5%, n=84/157), simplified referral paperwork (49.7%, n=78/157) and clearer pathways for onward management (44.6%, n=70/157).

We included a free-text question asking for suggestions to improve and develop genomic testing services to support better access. Comments were provided by 22 participants (14.0%). Inductive content analysis identified ten categories. The most frequently cited suggestions related to increasing the workforce to support offering genomic tests, for example, 'an increase in workforce proportionate to the work involved' (see online supplemental information III).

Education and training

Participants were asked to select from a list any prior general education in genetics and genomics they had had. The most frequent response was on-the-job education (49.0%, n=77/157), followed by hospital lectures (31.2%, n=49/157), self-directed learning (28.0%, n=44/157) and genetics seminars and workshops (21.0%, n=33/157) (table 5). Participants were also asked what education they access to keep up to date with genomic medicine. The largest proportion of respondents selected that they attended continuing professional development (CPD) meetings (n=46/157, 29.3%), however, 25.5% (n=40/157) reported not keeping up to date (table 5). Finally, participants were asked to select the three options they felt would be most useful in meeting their training needs (table 5). Webinars (48.4%, n=76/157), followed by CPD meetings and conferences (38.9%, n=61/157), were most frequently selected (table 5).

DISCUSSION

The NHS England report *Accelerating Genomic Medicine in the NHS* sets out the NHS's approach to "embed genomics across the NHS…from primary and community care through to specialist and tertiary care" as one of its four priority areas.[7] However, our findings from across the country show a mixed picture of mainstreaming. Specifically, our survey revealed that only around half (49.0%) of paediatricians felt prepared to use genomic sequencing tests in clinical practice despite close to 60% responding that they regularly offer either genetic and/or genomic tests and 75% reporting that they had requested genomic testing in the past 12 months. Over one-third reported that they asked another HCP to return results. Furthermore, our survey revealed that less than half of participants had experience in 8 or more of the 15 tasks identified as key components of delivering genomic testing in a mainstream setting, and clinician confidence was variable for many of these tasks. In fact, none of the genomic tasks listed in this survey achieved mean confidence scores at the upper end of the confidence scale, that is, 8 (out of 10) and above, and mean confidence was below 5 for 4 of

Table 5 Experiences and preferences for genomics education

	n (157)	%
Previous education in genetics and genomics		
On-the-job education (eg, clinical rounds, bedside teaching, case reviews)	77	49.0
Hospital-supported lectures on genetics or genomics (eg, grand rounds, continuing professional development (CPD) meetings)	49	31.2
Self-directed genetics or genomics education (eg, through journal articles, online course, moodle)	44	28.0
Seminar/workshop in genetics	33	21. 0
Genetics or genomics course in my initial professional training	20	12.7
MSc course in genetics or genomics	2	1.3
I have had no general education in genetics or genomics	21	13.4
I don't know	3	1.9
Other*	1	0.6
Keeping up to date with genomic medicine		
CPD meetings	46	29.3
Webinars/face-to-face training sessions delivered by a Royal College or other professional body	36	22.9
Participating in multidisciplinary meetings	33	21.0
Guidelines from professional bodies	28	17.8
Journal articles	25	15.9
Training from local geneticist/genetic counsellor	24	15.3
National genomic sequencing educational MDT workshops	18	11.5
Local genomic medicine service/genomic laboratory website	16	10.2
External genetic or genomic seminars or conferences	14	8.9
Emails from NHS Genomics or the testing laboratory	13	8.3
Internal genetic or genomic seminars	7	4.5
Variant interpretation educational workshops	3	1.9
I don't currently keep updated about genetics or genomics	40	25.5
Other†	4	2.5
Preferences for training		
Webinar	76	48.4
CPD meetings and/or conferences	61	38.9
Face-to-face training	57	36.3
Self-paced e-learning course/module	55	35.0
Group case discussion/reflection	51	32.5
Regional sequencing educational MDT workshops	44	28.0
Written information	28	17.8
More frequent in-person or small-group variant interpretation educational workshops	24	15.3
Hands-on learning	23	14.6
More frequent online national sequencing educational MDT workshops	21	13.4
Inclusion in mandatory training at your hospital	18	11.5
Other†	2	1.3

*Other training attended included research (n=1).
†No alternatives were specified.
MDT, multidisciplinary team; NHS, National Health Service.

the 15 genomic tasks listed, including three tasks related to returning genomic results to patients.

Our study design has notable strengths. We recruited participants within a short time period (2 months) providing us with a snapshot of paediatricians' experiences with WGS around 3 years since the mainstreaming agenda was introduced. In addition, we successfully recruited 157 participants, which provided a sufficient

number of responses to conduct meaningful analyses and draw preliminary insights. Our sample was, in certain aspects, representative of the study population. For example, majority female, in their late 40s, consultants and hospital-based, which is in line with a recent workforce census report from the RCPCH (Royal College of Paediatrics and Child Health).[26] However, our recruitment methods, particularly recruiting through a conference and through membership of professional organisations, may have biased the sample to more professionally engaged clinicians and those more likely to engage in CPD activities. As such, our findings may not be generalisable as these participants may be more well informed about the use of genomic technologies and more confident accessing genomic testing. Questions were not filtered using question logic in our survey. Therefore, in some instances, participants answered 'no' to a question but answered the next question as if they had answered 'yes', potentially impacting the validity of the results. However, the numbers were small and did not significantly change the interpretation of the results in those cases. Furthermore, given the nature of our recruitment methods, we are unable to calculate a response rate for the survey, and we were unable to check for multiple participation of participants. Finally, some terms/definitions were open to interpretation, for example, what CPD consists of. Participants may have interpreted these terms differently.

Even though we found preparedness and confidence among paediatricians in England to be low, they appear higher than in studies conducted in other countries with a mainstreaming agenda (although comparing findings across studies needs to be done with caution given that the measures used varied). Only a quarter of health professionals from across 30 different medical specialties surveyed in Australia reported feeling sufficiently prepared to use WGS testing in their clinical practice. Only one-third of non-genetic clinicians and primary care providers offering testing in the USA reported feeling sufficiently prepared, and less than half of neurologists surveyed worldwide reported feeling sufficiently prepared.[9 12 27] The findings from our study may be due to the 'top-down' national approach to implementation of WGS that took place in England, which followed on from the 100 000 Genomes Project, which was essentially a pilot study for providing genome sequencing in a national healthcare system.[28] This differs from the approaches in Australia and the USA where implementation was 'bottom-up' whereby genomic sequencing was implemented via a piecemeal, state-by-state approach.[28] Moreover, they may reflect prioritisation of the mainstreaming agenda by the UK government and the various education and training opportunities set up by NHS England and Health Education England, including a Master's in Genomic Medicine, online courses, interactive sessions and teaching materials.[7 8 29 30]

In this study, just under half of participants reported returning genomic results to patients and families, and of those, responders were moderately confident discussing straightforward results and had low confidence discussing more complex results. Paediatricians should at a minimum be confident to return straightforward results to their patients so that clinical geneticists can be freed up to focus on more complex results. Increasing access to 'just-in-time' resources such as GeNotes (online quick-access concise information to support HCPs' genomic decision-making), as well as having MDT meetings to facilitate the discussion of complex cases, is likely to improve paediatrician confidence over time.[31] In fact, each NHS Genomic Laboratory Hub has been funded to support genomic MDT meetings, and regional specialty-specific MDT meetings have been established.[6] Interviews conducted as part of this broader, mixed-methods study revealed that community paediatricians in England were found to find routinely organised MDT meetings beneficial for up-skilling, and around half of participants had attended MDT meetings with clinical colleagues.[16] Reasons as to why some paediatricians are not attending MDT meetings are unclear, and further research here would be beneficial to understand if the barrier relates to lack of awareness, perceived utility or other reasons.

Another notable finding relating to returning genomic test results was that nearly a quarter of participants who had ordered a test in the past 12 months had not returned results as they had not yet received them. This is likely to reflect the long turnaround times currently being experienced in the NHS in reporting genomic results. NHS England has established guidelines for reporting times based on the urgency and complexity of genomic tests. For large gene panels or WGS (non-urgent, complex tests), the target is 84 calendar days (12 weeks).[32] However, data show that few results are returned within this target. For example, recent data from the Cambridge laboratory report that only 13% of results are returned within this timeframe.[33] Findings from another study within this mixed-methods evaluation highlighted a number of barriers to timely reporting of results, including reliance on paper form-filling, lack of clinical scientists and poorly coordinated IT systems across the country as key factors associated with the significant backlog.[34]

This study found that genomic confidence was significantly higher among those who regularly offer testing compared with those who do not. These results are not surprising; learning by doing is an active learning methodology which goes back to Aristotle who said, "What we have to learn, we learn by doing". McClaren *et al* came to a similar conclusion after they set out to explore the continuing education needs of a wide range of non-genetic specialists from across Australia.[35] Their findings showed that participants believed that experiential learning, including learning by observation, was necessary to develop the confidence and skills needed for clinical care.[35]

Even though only 16% of participants thought that 'genomic champions' would help support them in offering genomic testing in this study, which may

potentially be because their role among paediatricians is unclear, their importance in supporting the mainstreaming agenda through imparting knowledge and best practice has been noted by genomic experts in the broader study associated with this work.[34] In the GMS, 'genomic champions' have been created across a range of clinical specialties to advocate and promote the use of genomic medicine within their organisation or community. These champions play a pivotal role in integrating genomics into mainstream settings by raising awareness, promoting education and training and providing guidance on genomic testing. Funding and promoting the role of genomic champions as well as having genomic associates and/or genetic counsellors embedded within paediatric settings may help to drive the implementation of genomic testing and improve paediatrician preparedness and confidence.[34] Given that paediatricians reported finding the consenting process complex and time-consuming, genomic associates in particular could reduce some of their workload.

The preferred training provision in this survey was through webinars and CPD meetings and/or conferences. Increased genomic CPD has been shown to increase confidence and increase referrals for testing.[36] Nisselle *et al* reported participants having a strong interest in further genomics education and, as with this study, prefer it incorporated into their normal workplace activities including CPD activities and workplace seminars as well as learning from peers.[9] Targeting workplace training, perhaps as part of local CPD seminar programmes, to support clinicians' understanding of genomic information and how to incorporate results into patient management is likely to improve both capability and motivation for mainstreaming.

CONCLUSIONS

Paediatricians are expected to deliver genomic medicine as part of the NHS's mainstreaming agenda. Our study provides a snapshot into how well prepared and confident this specialist group feels during the early years of the GMS and suggests there is still work to be done. Further research with other non-genetic specialists would provide a more complete picture of how confident and prepared the workforce feels to integrate genomic medicine across the health system from primary to tertiary care. Our study could also be repeated at a future time point to see whether confidence and preparedness have increased over time.

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Patient consent for publication Not applicable.

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